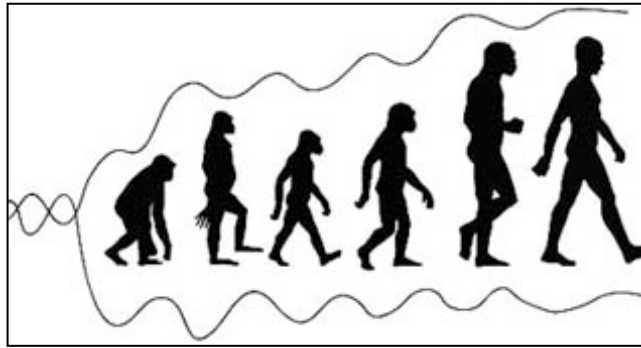


Y chromosome research: past, present and future



Chris Tyler-Smith

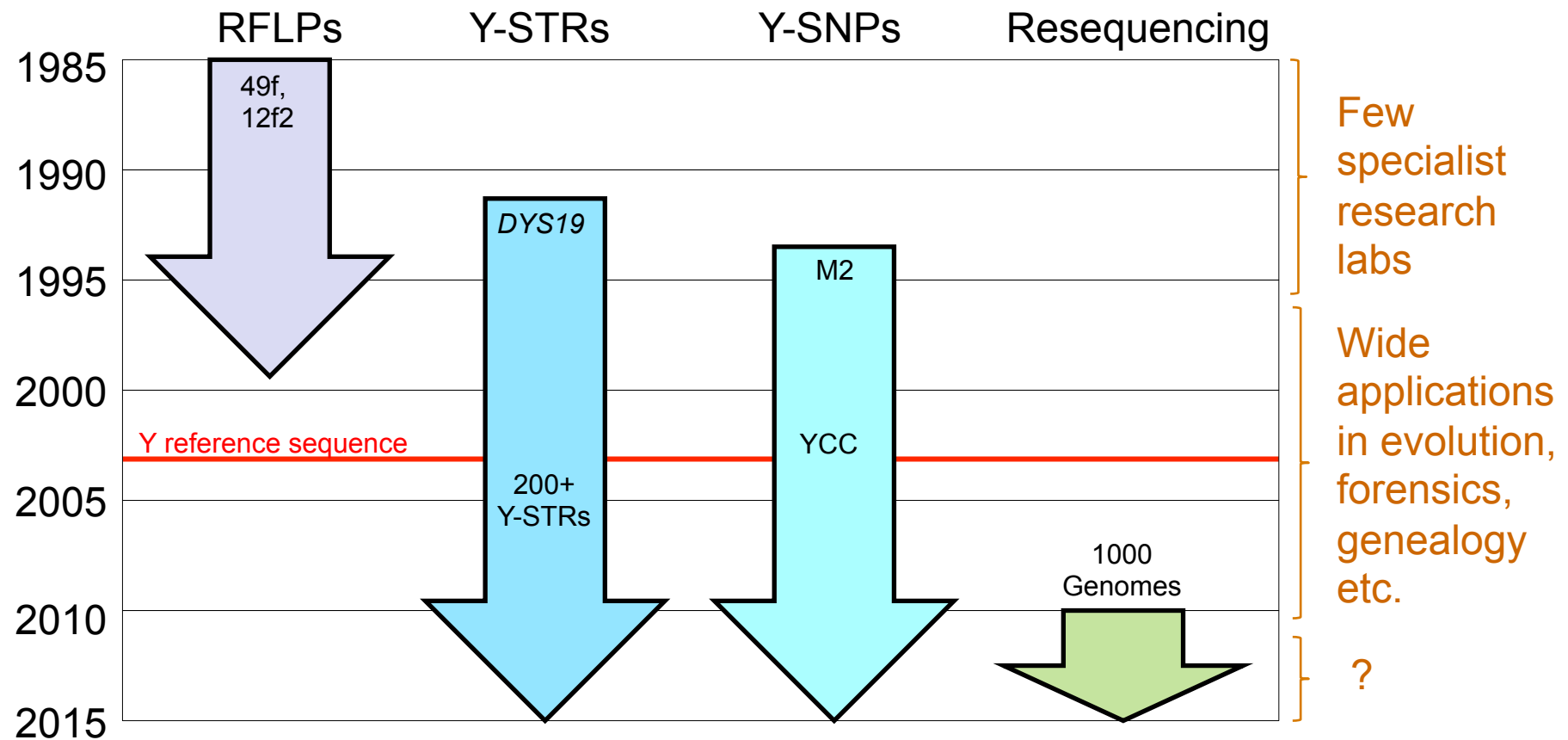
The Wellcome Trust Sanger Institute, Hinxton, UK



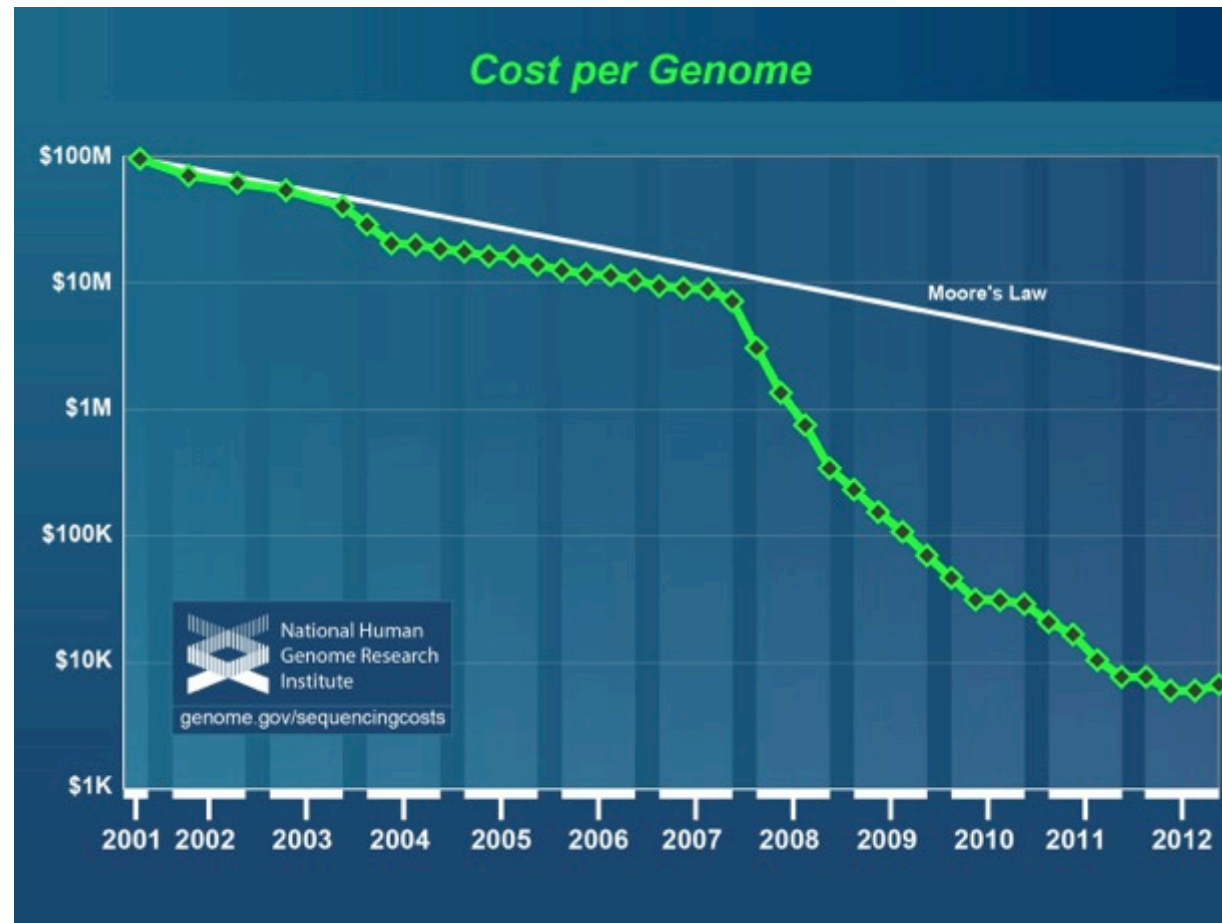
Summary

- Past
 - From RFLPs to Y-SNPs + Y-STRs to resequencing
- Present
 - Large-scale sequencing and genotyping
- Future
 - Issues for the field

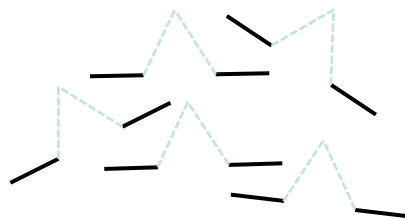
History of Y chromosome research



Sequencing costs

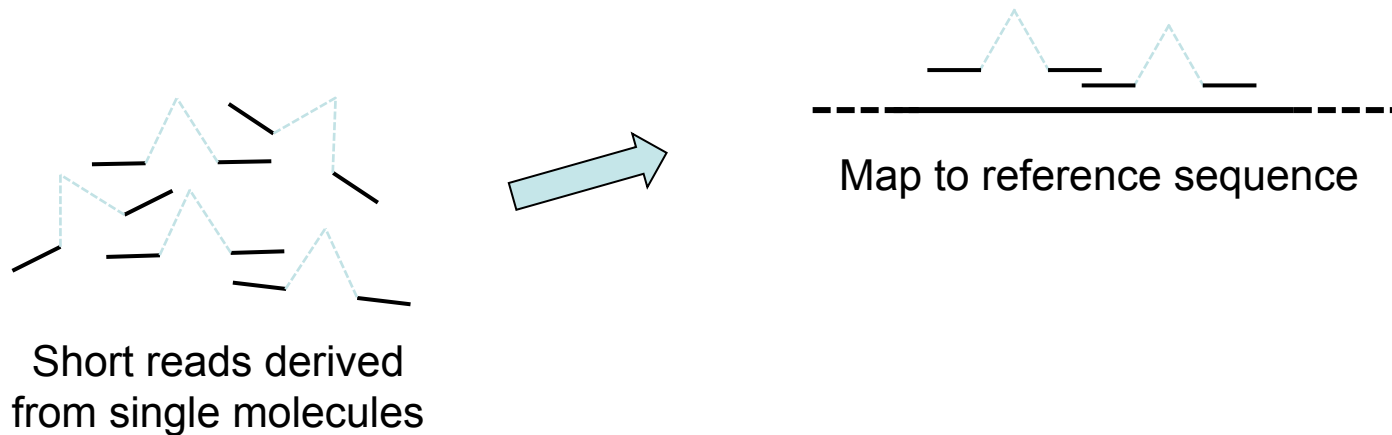


Next-Gen Sequencing: steps

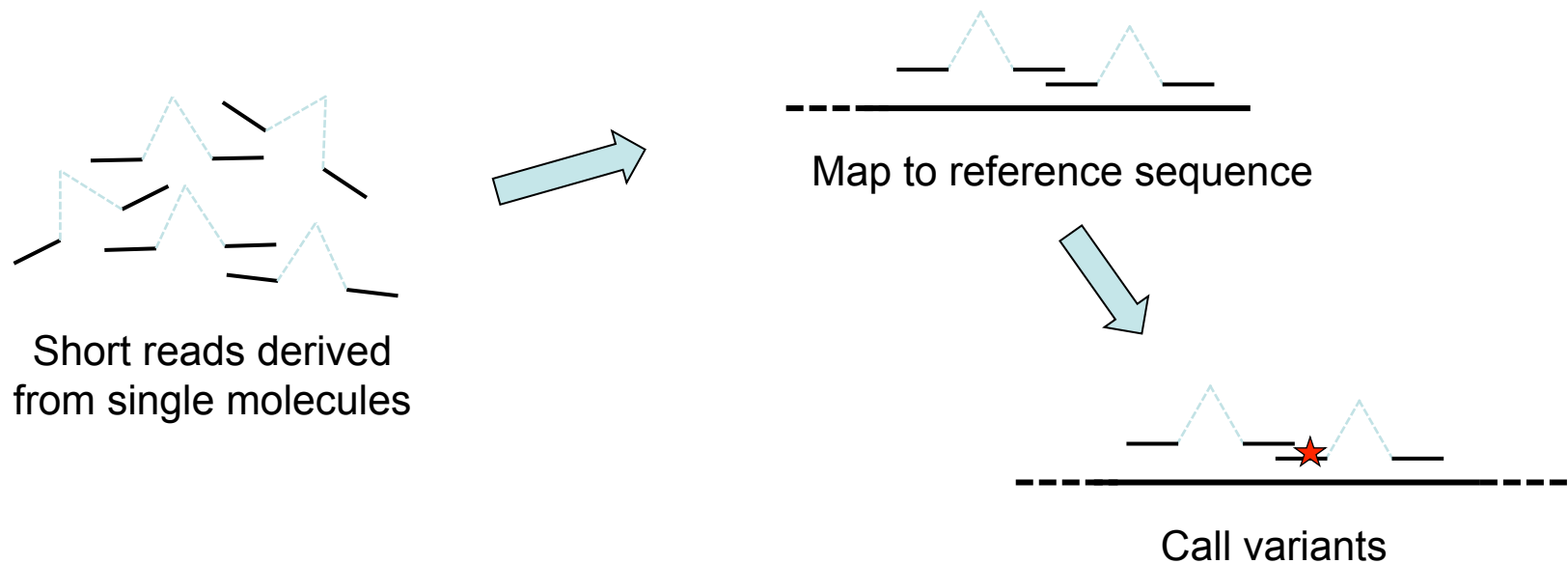


Short reads derived
from single molecules

Next-Gen Sequencing: steps

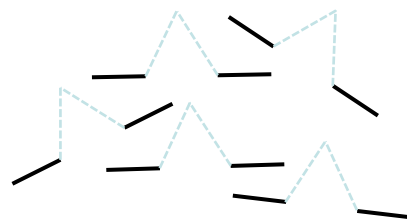


Next-Gen Sequencing: steps



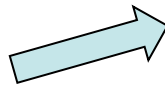
Next-Gen Sequencing: steps

Base-calling errors



Short reads derived from single molecules

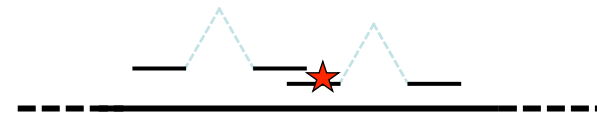
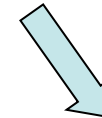
Duplicate reads



Mapping errors



Map to reference sequence

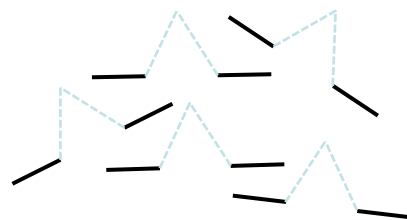


Call variants

Variant-calling errors

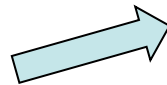
Next-Gen Sequencing: steps

Base-calling errors

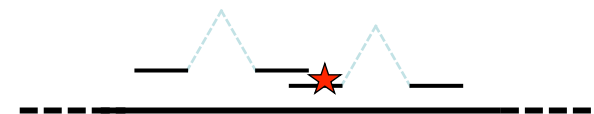


Short reads derived from single molecules

Duplicate reads

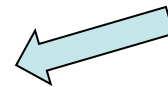


Map to reference sequence



Call variants

Filter and Validate



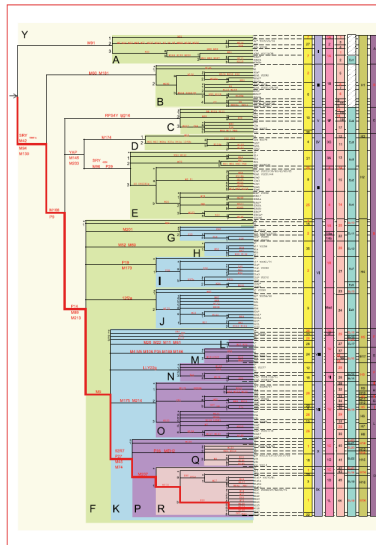
Y chromosome sequence datasets are becoming available

OPEN ACCESS Freely available online

PLOS BIOLOGY

The Diploid Genome Sequence of an Individual Human

Samuel Levy^{1*}, Granger Sutton¹, Pauline C. Ng¹, Lars Feuk², Aaron L. Halpern¹, Brian P. Walenz¹, Nelson Axelrod¹, Jiaqi Huang¹, Ewen F. Kirkness¹, Gennady Denisov¹, Yuan Lin¹, Jeffrey R. MacDonald², Andy Wing Chun Pang², Mary Shago², Timothy B. Stockwell¹, Alexia Tsimouri¹, Vineet Bafna³, Vikas Bansal², Saul A. Kravitz¹, Dana A. Busam¹, Karen Y. Beeson¹, Tina C. McIntosh¹, Karin A. Remington¹, Josep F. Abril⁴, John Gill¹, Jon Borman¹, Yu-Hui Rogers¹, Marvin E. Frazier¹, Stephen W. Scherer², Robert L. Strausberg¹, J. Craig Venter¹



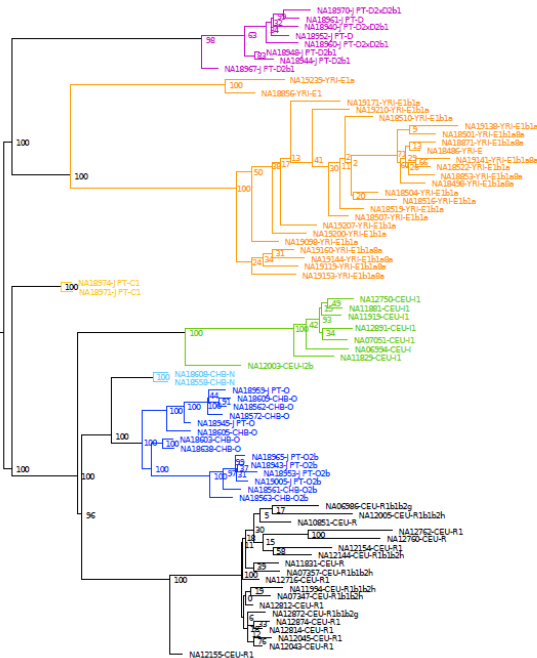
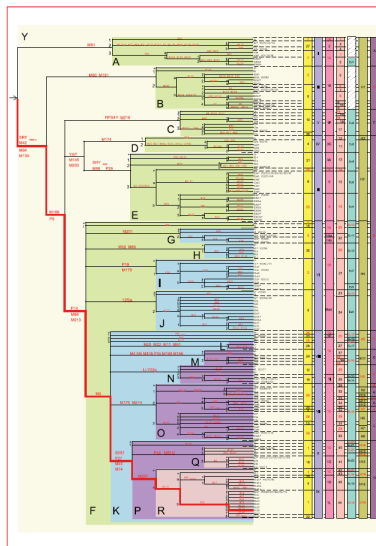
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ARTICLE

doi:10.1371/journal.pbio.1001544

A map of human genome variation from population-scale sequencing

The 1000 Genomes Project Consortium*

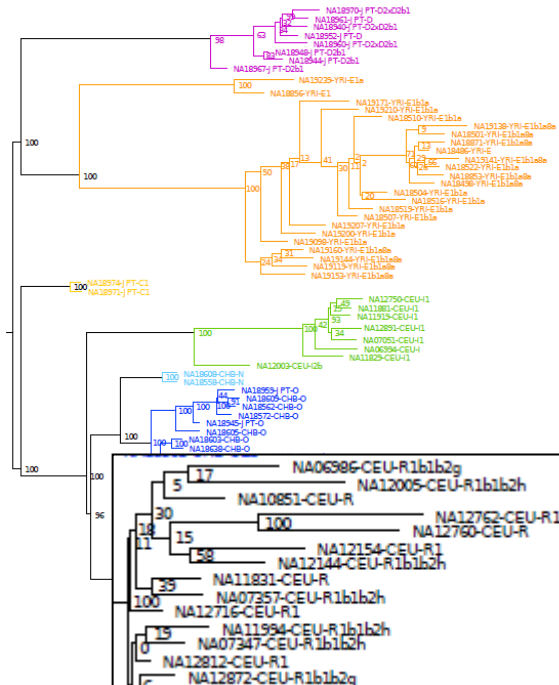
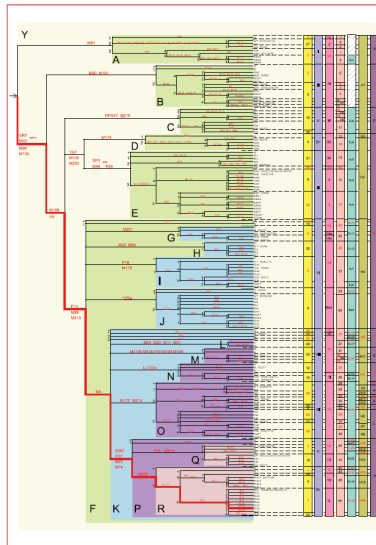
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ARTICLE

doi:10.1038/nature09533

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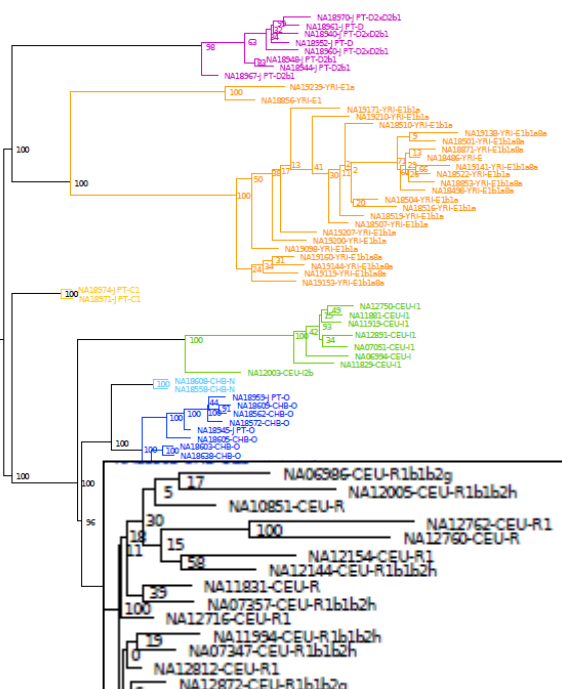
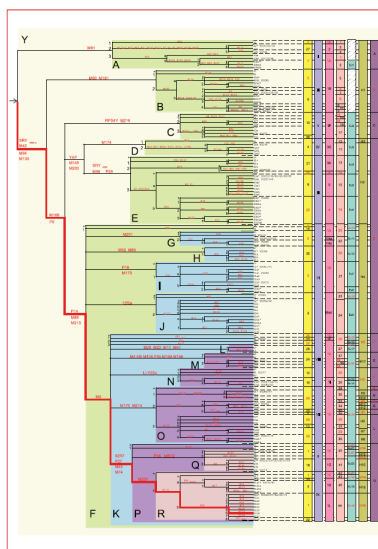
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doi:10.1016/j.neuroimage.2016.03.032

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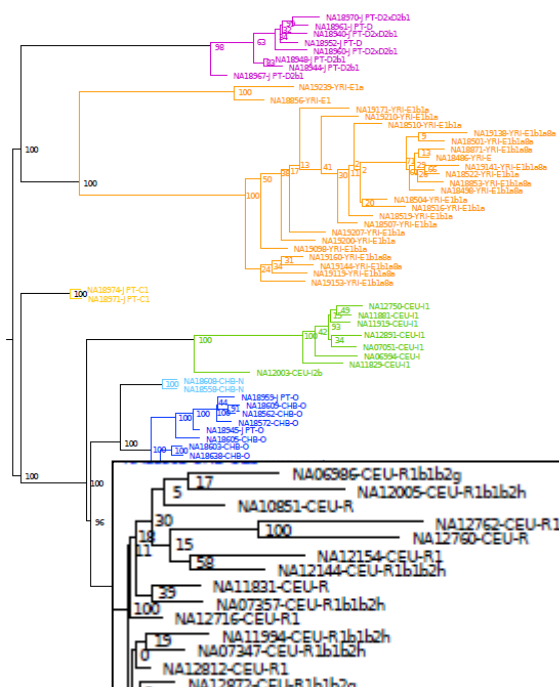
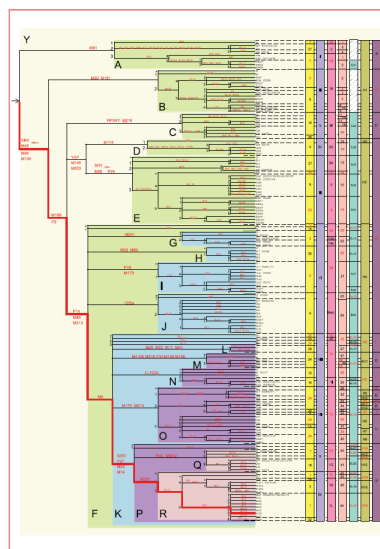
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ARTICLE

doi:10.1038/nature11632

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Resource:

A calibrated human Y-chromosomal phylogeny based on resequencing

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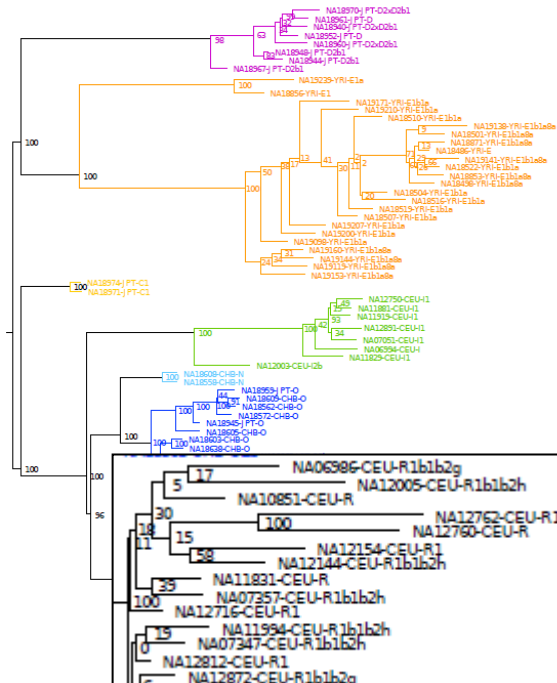
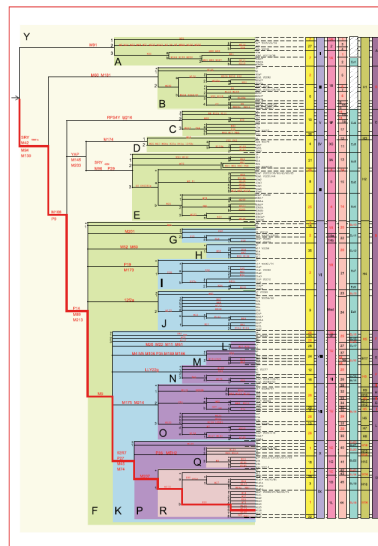
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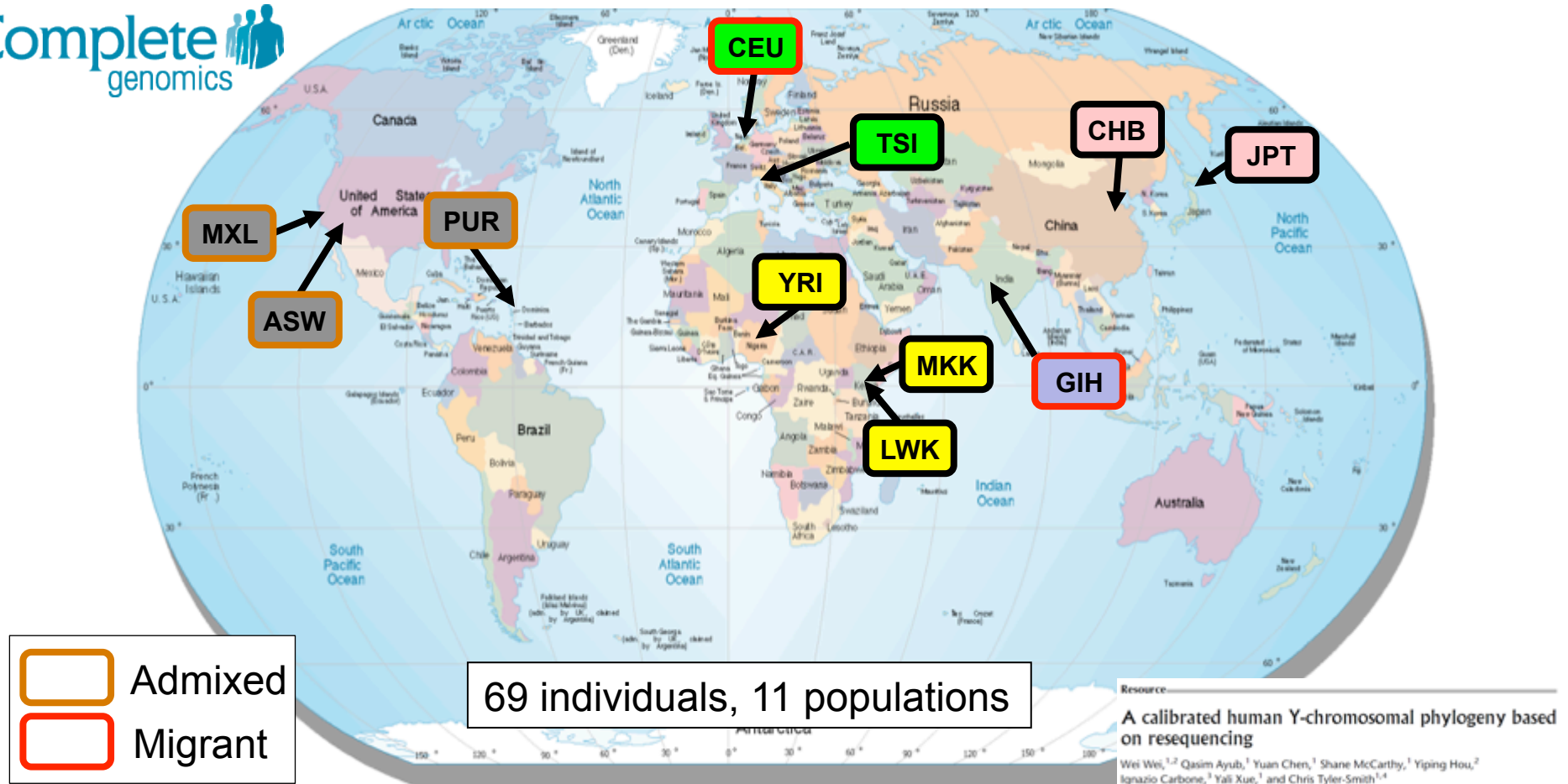
ARTICLE

Deep Whole-Genome Sequencing of 100 Southeast Asian Malays

Lai-Ping Wong,^{1,14} Rick Twee-Hee Ong,^{1,14} Wan-Ting Poh,^{1,14} Xuanyao Liu,^{1,2,14} Peng Chen,¹ Ruoying Li,¹ Kevin Koi-Yau Lam,¹ Nisha Esakimuthu Pillai,³ Kar-Seng Sim,⁴ Halyan Xu,¹ Ngak-Leng Sim,⁴ Shu-Mei Teo,^{1,2} Jia-Nee Foo,⁴ Linda Wei-Lin Tan,¹ Yenly Lim,¹ Seok-Hwee Koo,⁵ Linda Seo-Hwee Gan,⁶ Ching-Yu Cheng,^{1,10,11} Sharon Wee,¹ Eric Peng-Huat Yap,⁶ Pauline Crystal Ng,⁴ Wei-Yen Lim,¹ Richie Soong,⁷ Markus Rene Wenk,^{8,9} Tin Aung,^{10,11} Tien-Yin Wong,^{10,11} Chiea-Chuen Khor,^{1,4,10,12} Peter Little,³ Kee-Seng Chia,¹ and Yik-Yang Teo^{1,2,3,4,13*}

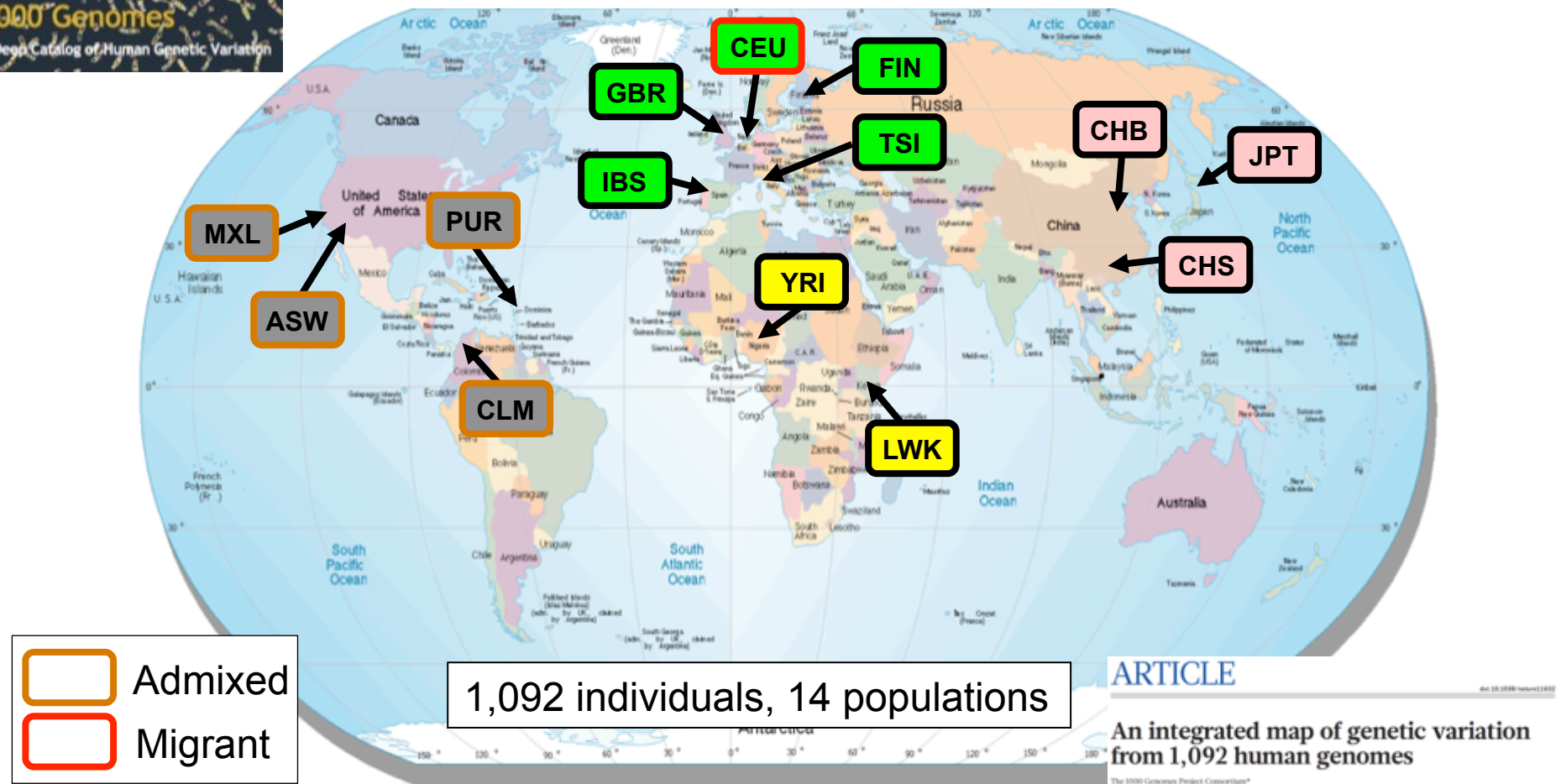
Recent and current research

36 high-coverage Y chromosomes



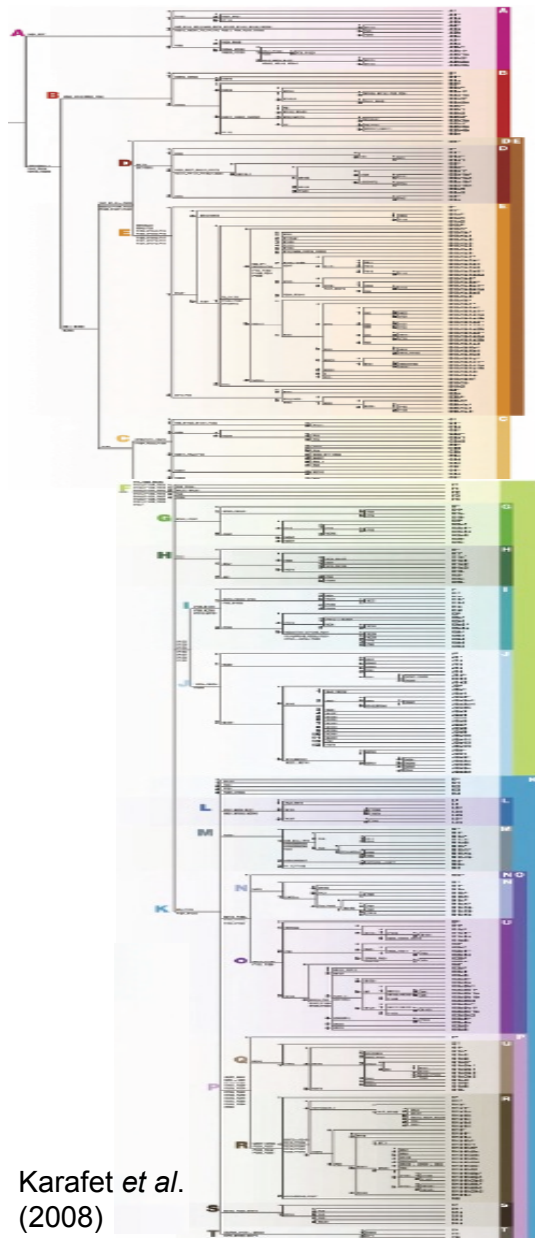
523 low-coverage Y chromosomes

1000 Genomes
A Data Catalog of Human Genetic Variation

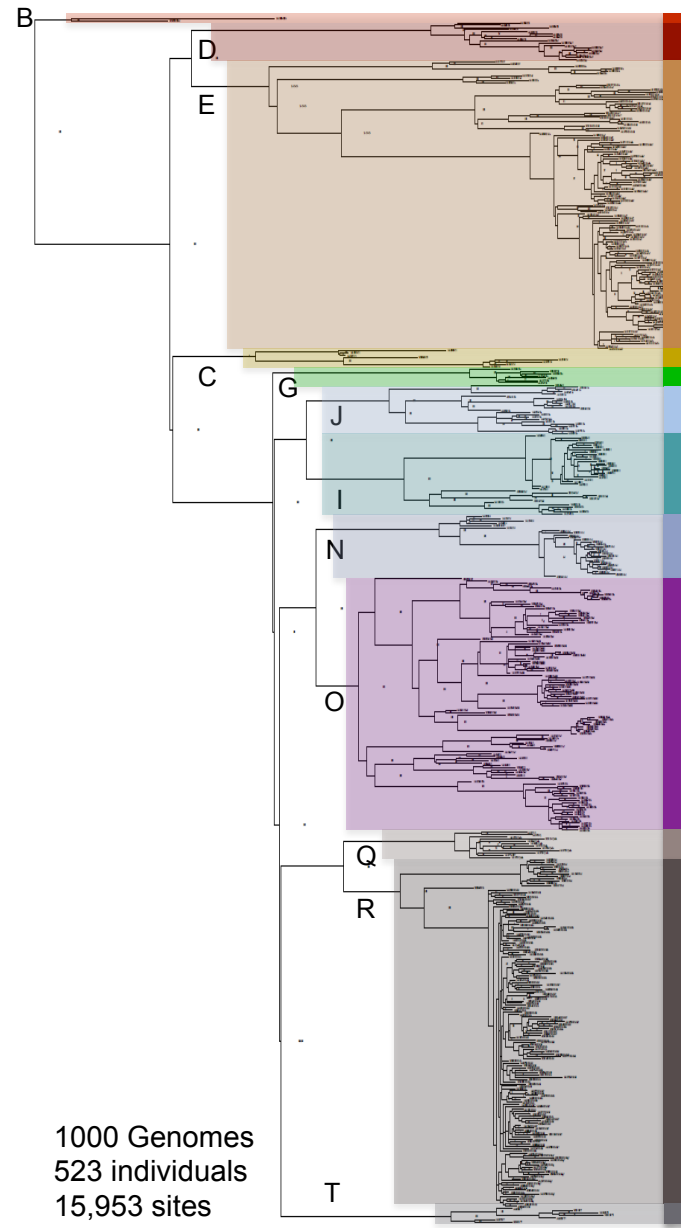
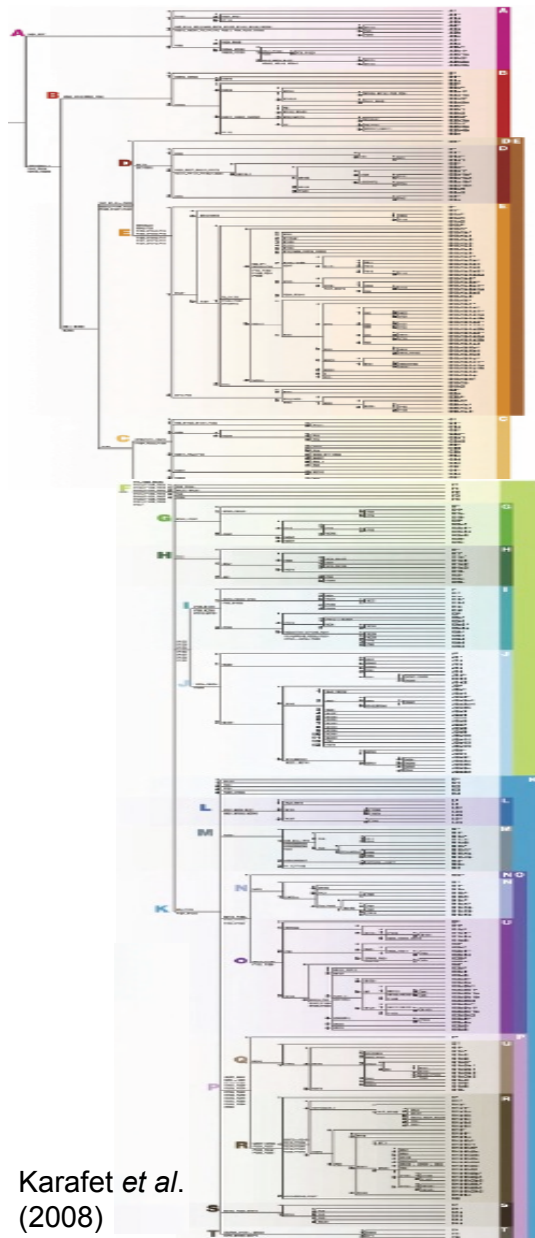


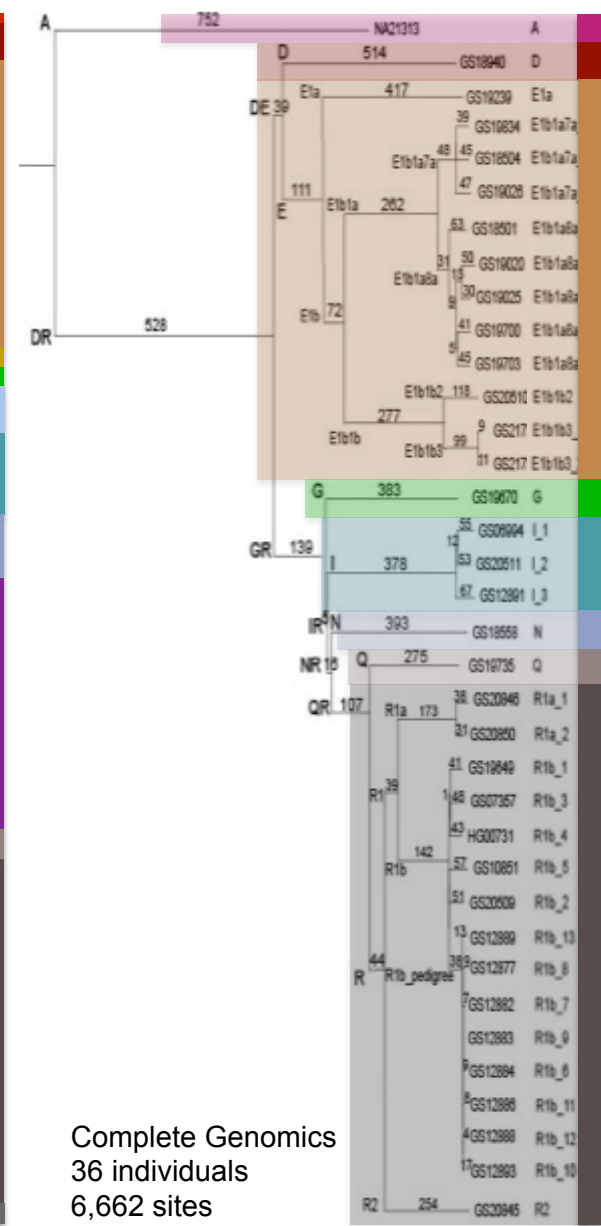
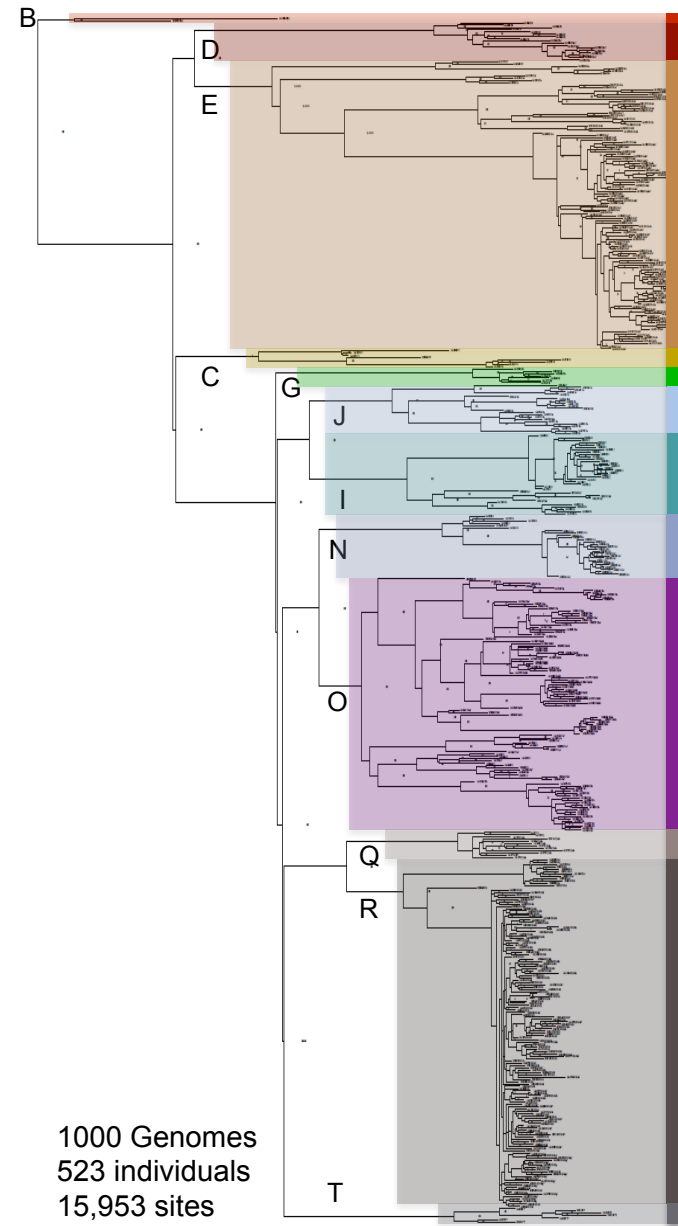
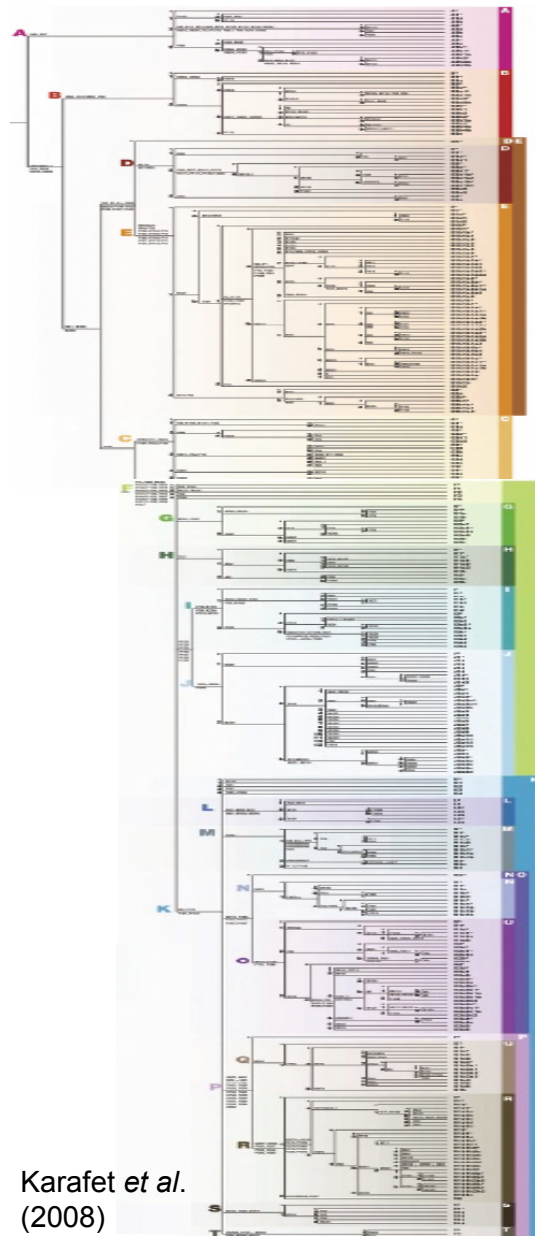
What have we discovered from sequencing lots of Y chromosomes?

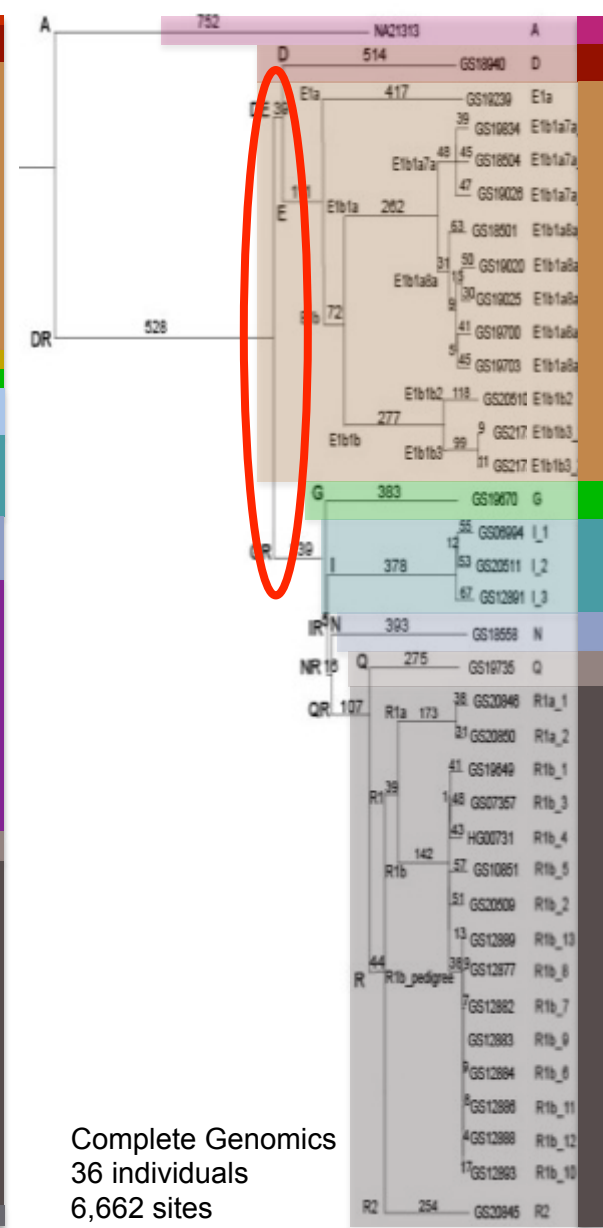
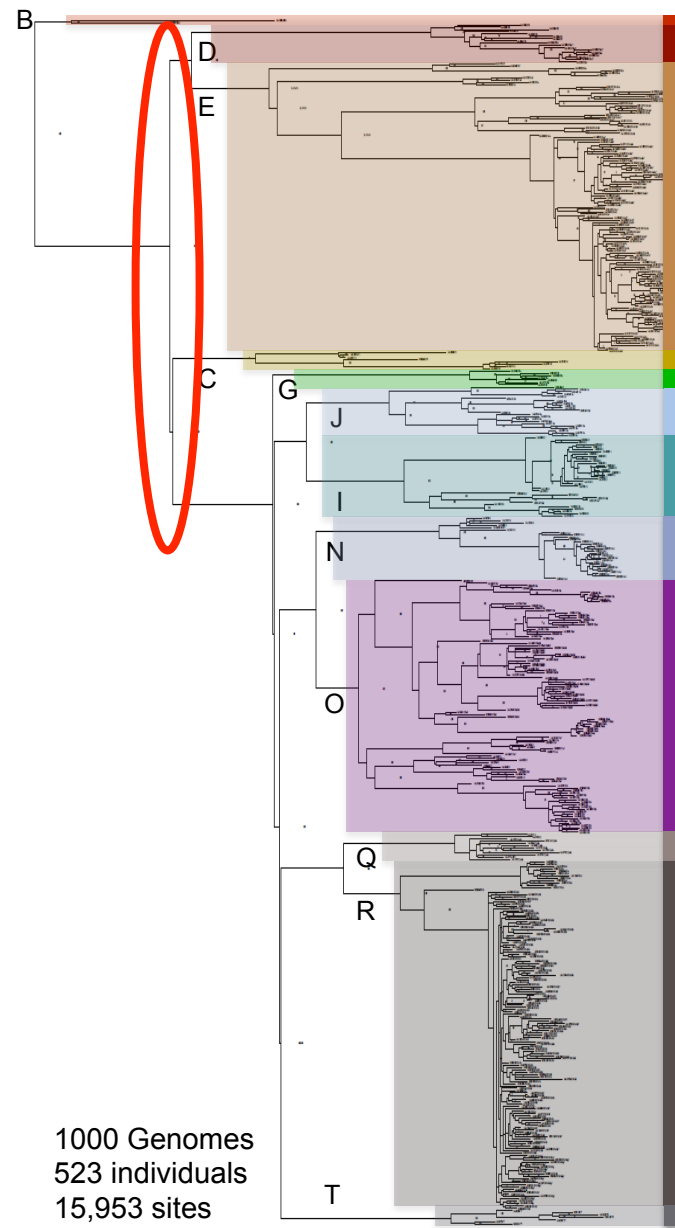
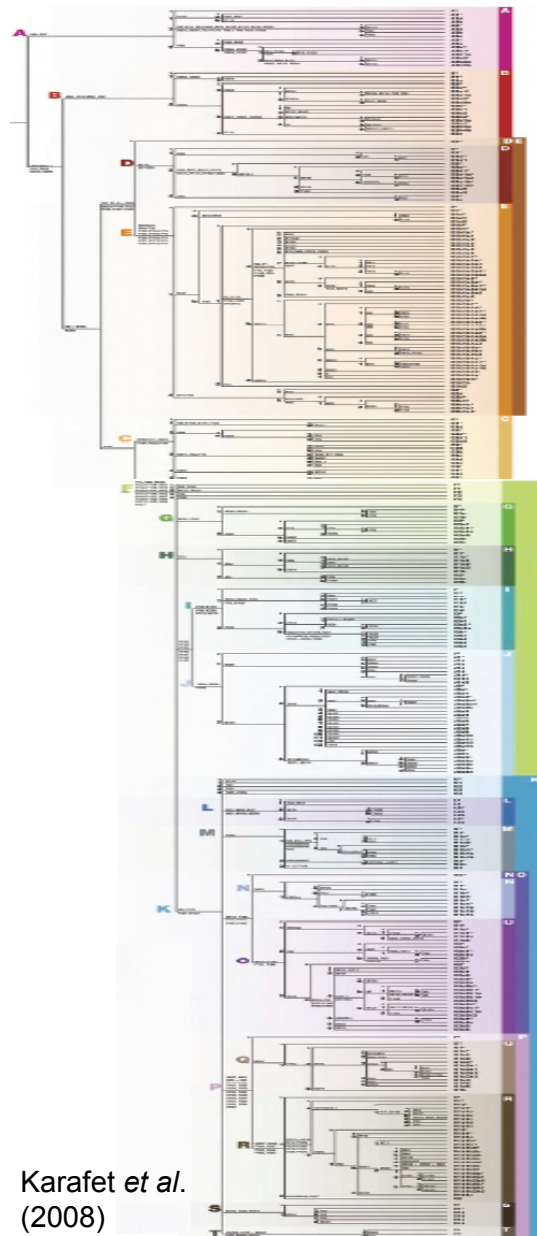
- Lots of SNPs and other variants
 - 36 Y chrs: 6,662 variants
 - 523 Y chrs: 15,953 variants
- The structure of the Y phylogeny
- The timescale of the Y phylogeny

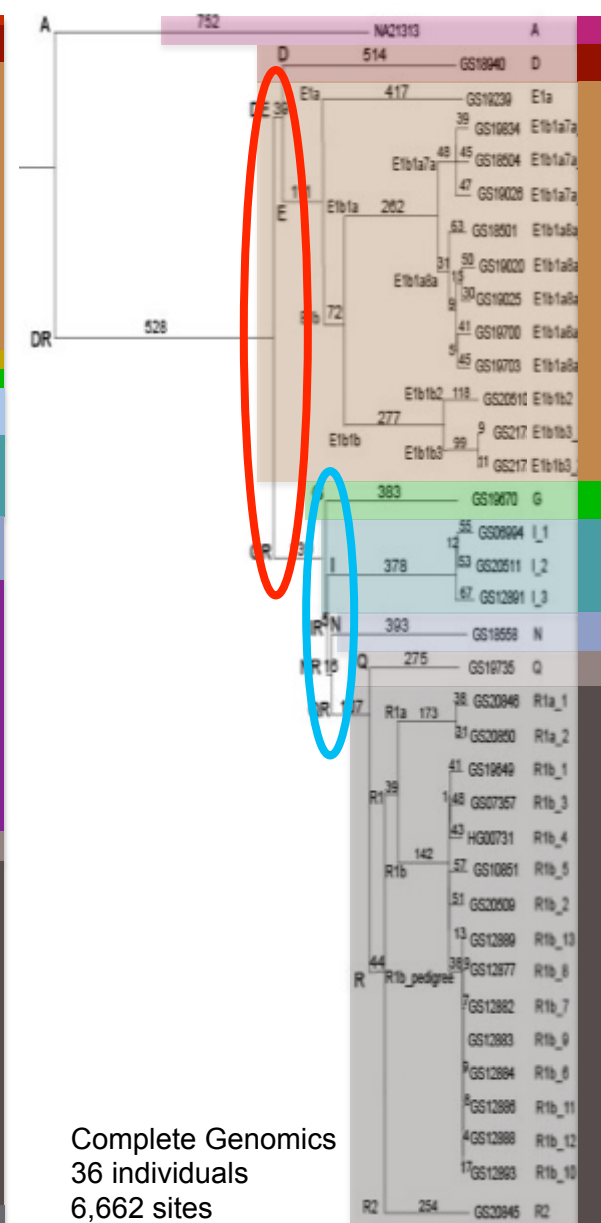
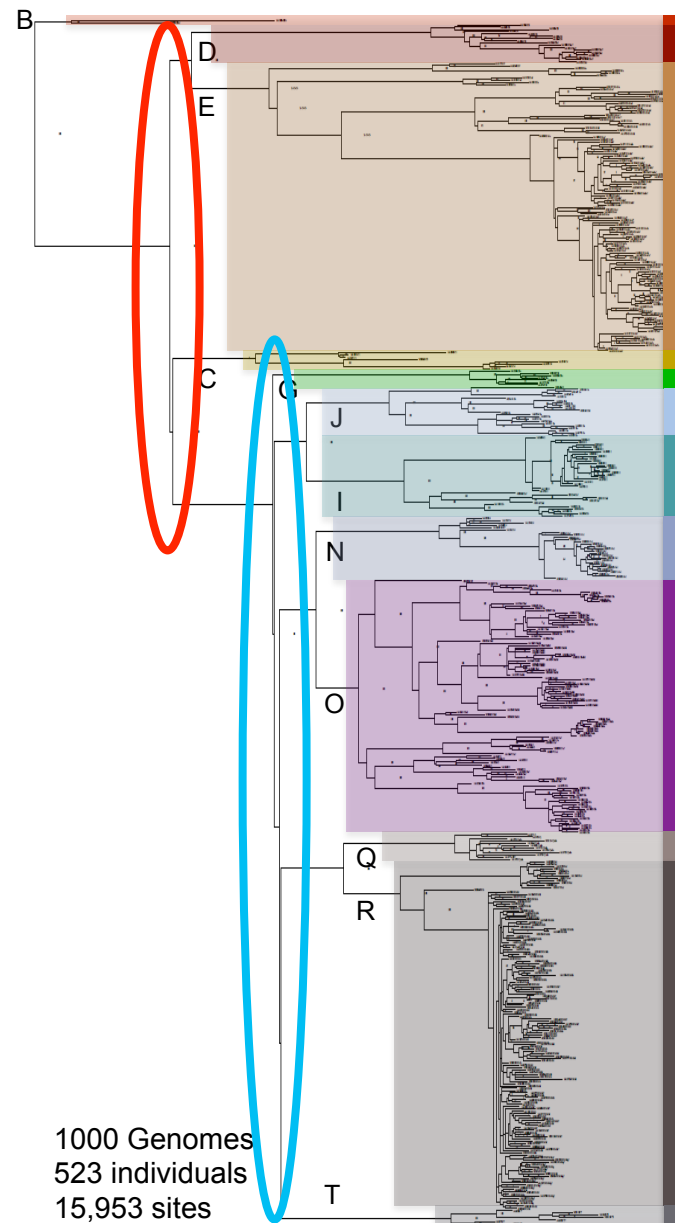
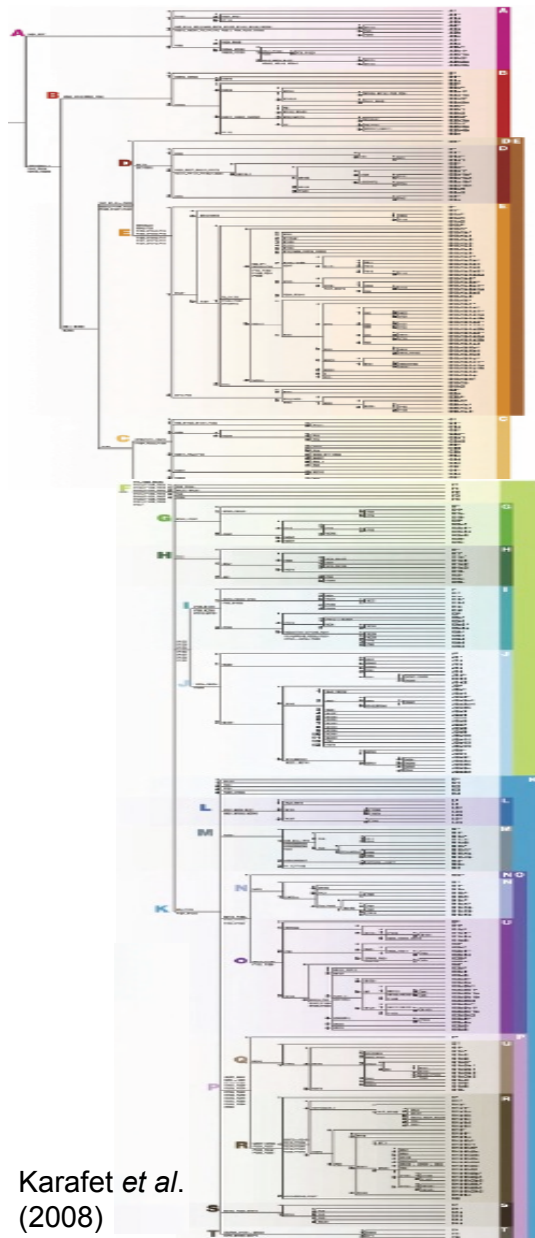


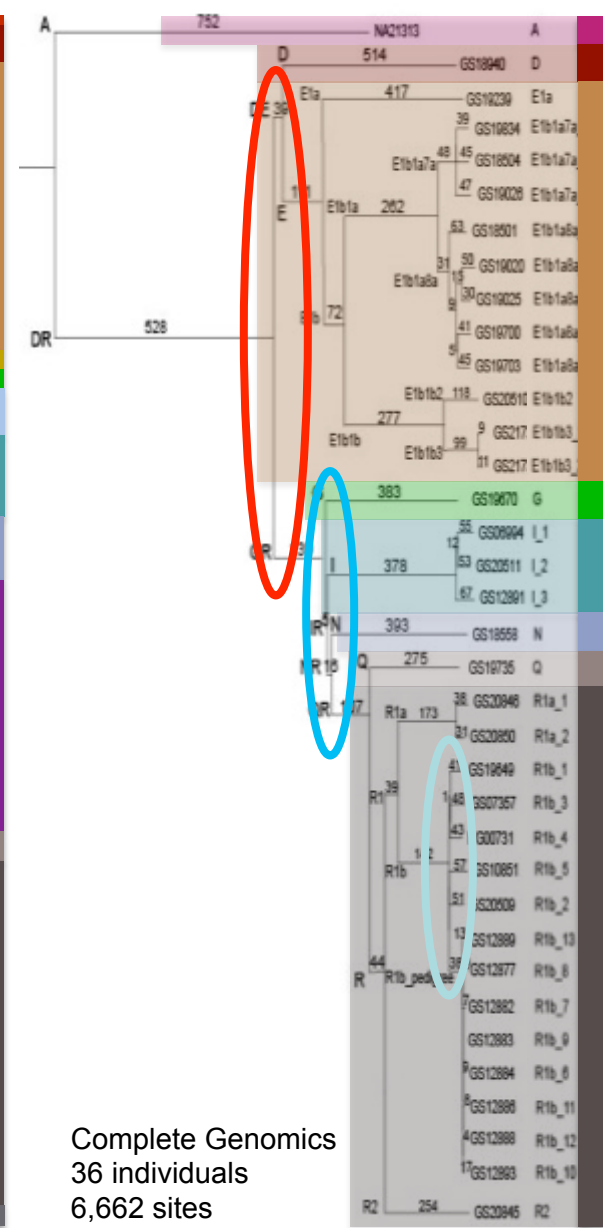
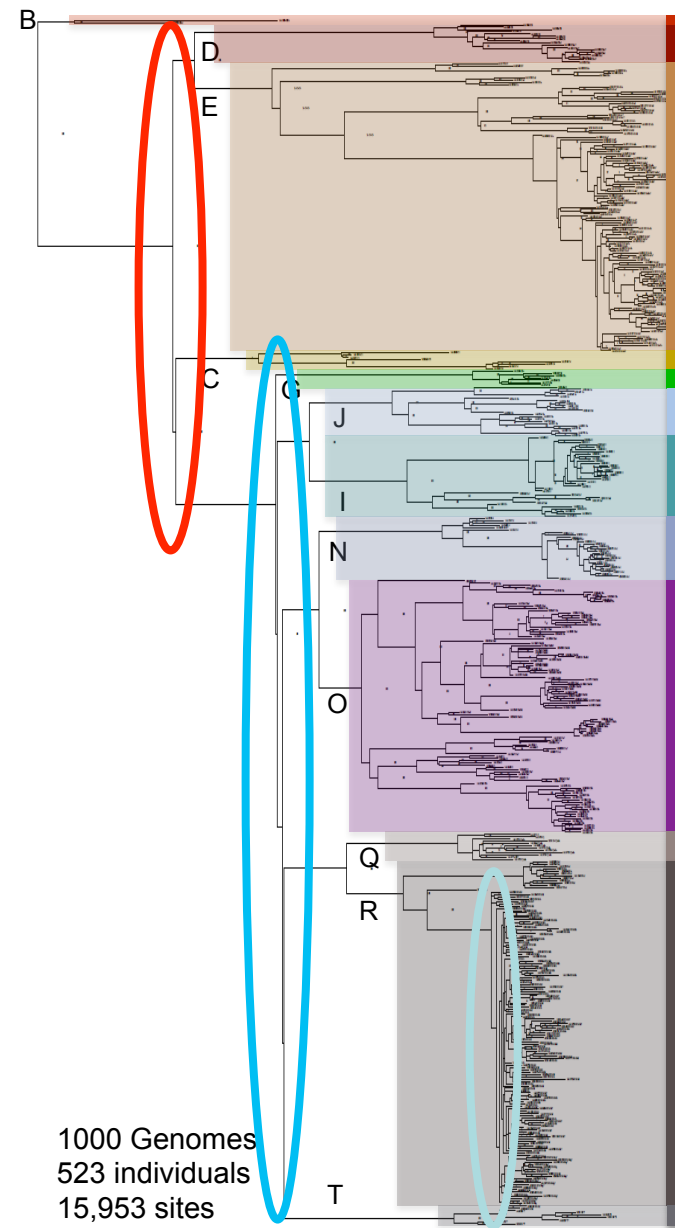
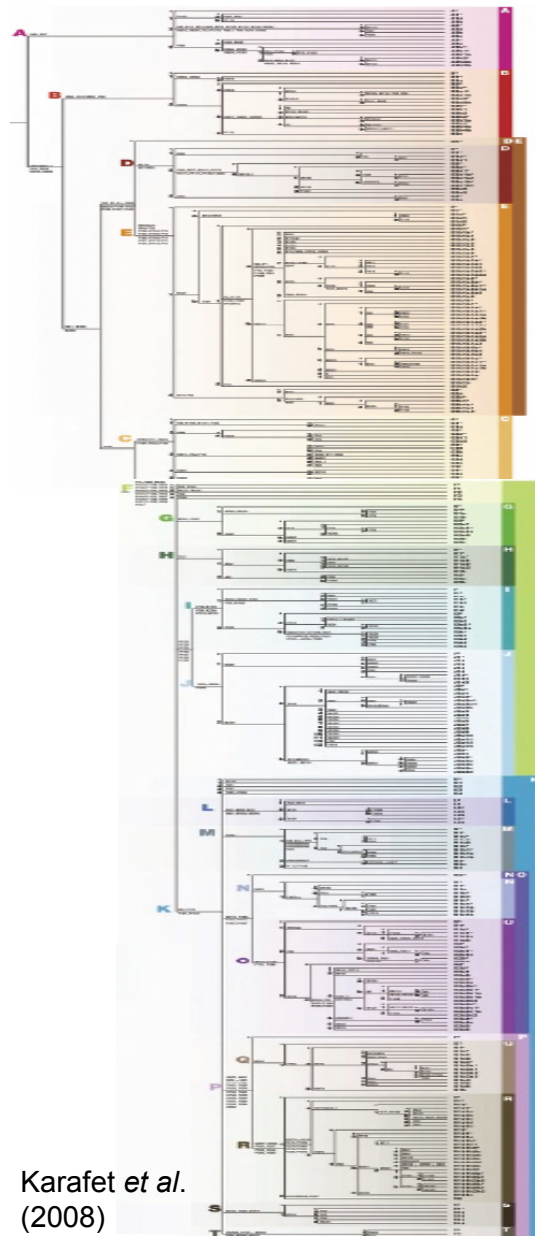
Karafet *et al.*
(2008)





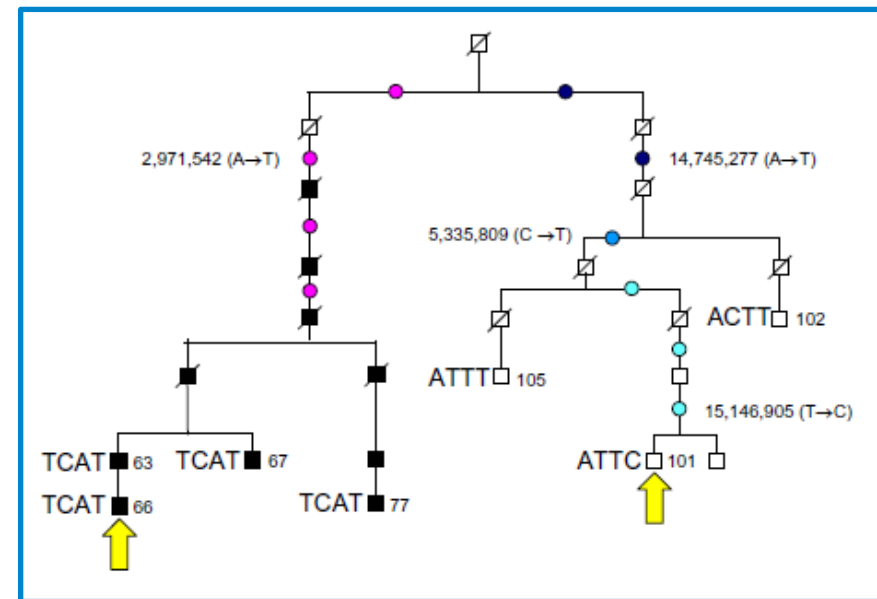






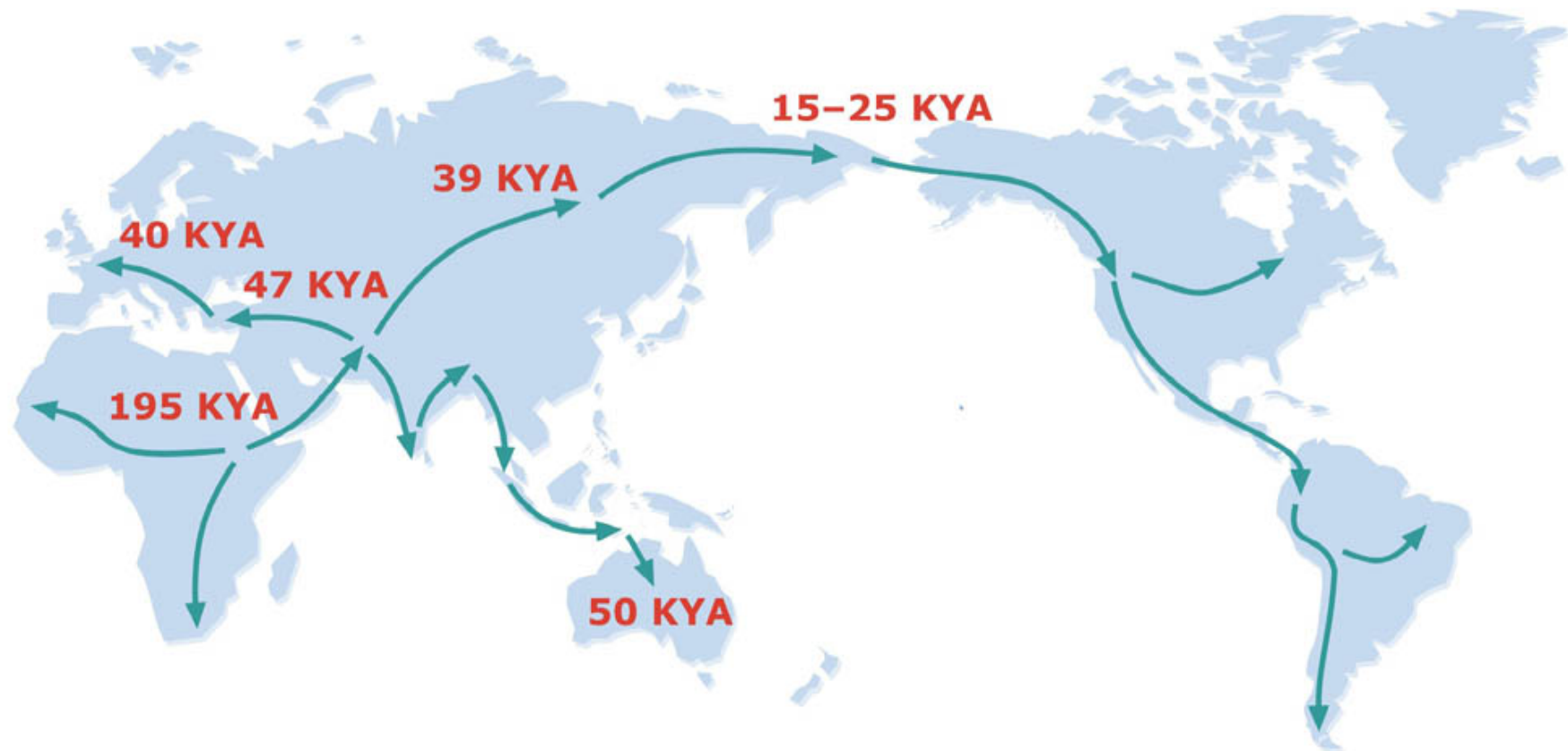
Calibrating the Y-SNP mutation rate

- Sequence two Y chromosomes from an extended family
- 4 mutations in 10.15 Mb
- Mutation rate 3.0×10^{-8} /nucleotide/generation (95% CI: 8.9×10^{-9} – 7.0×10^{-8})

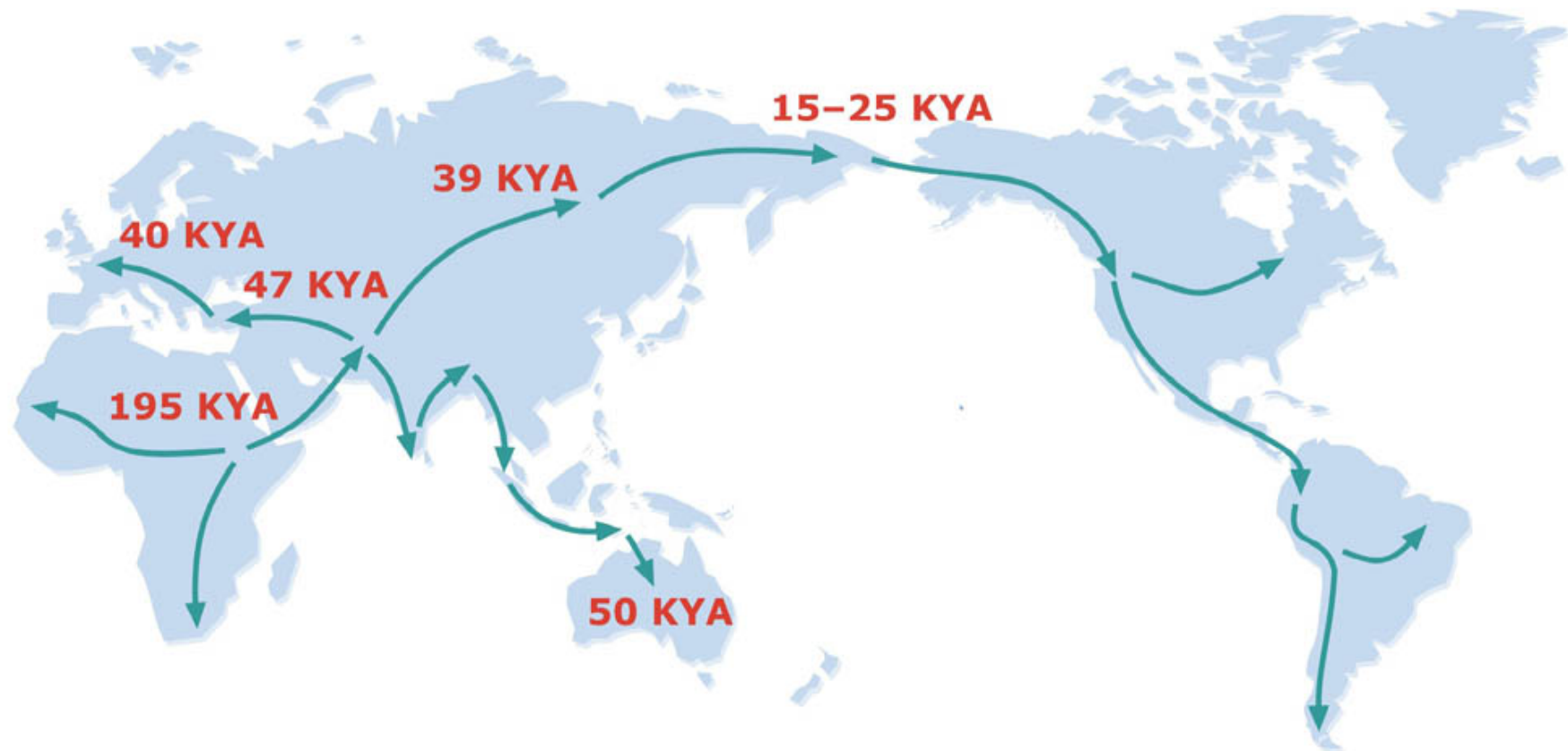


13 generations
↔

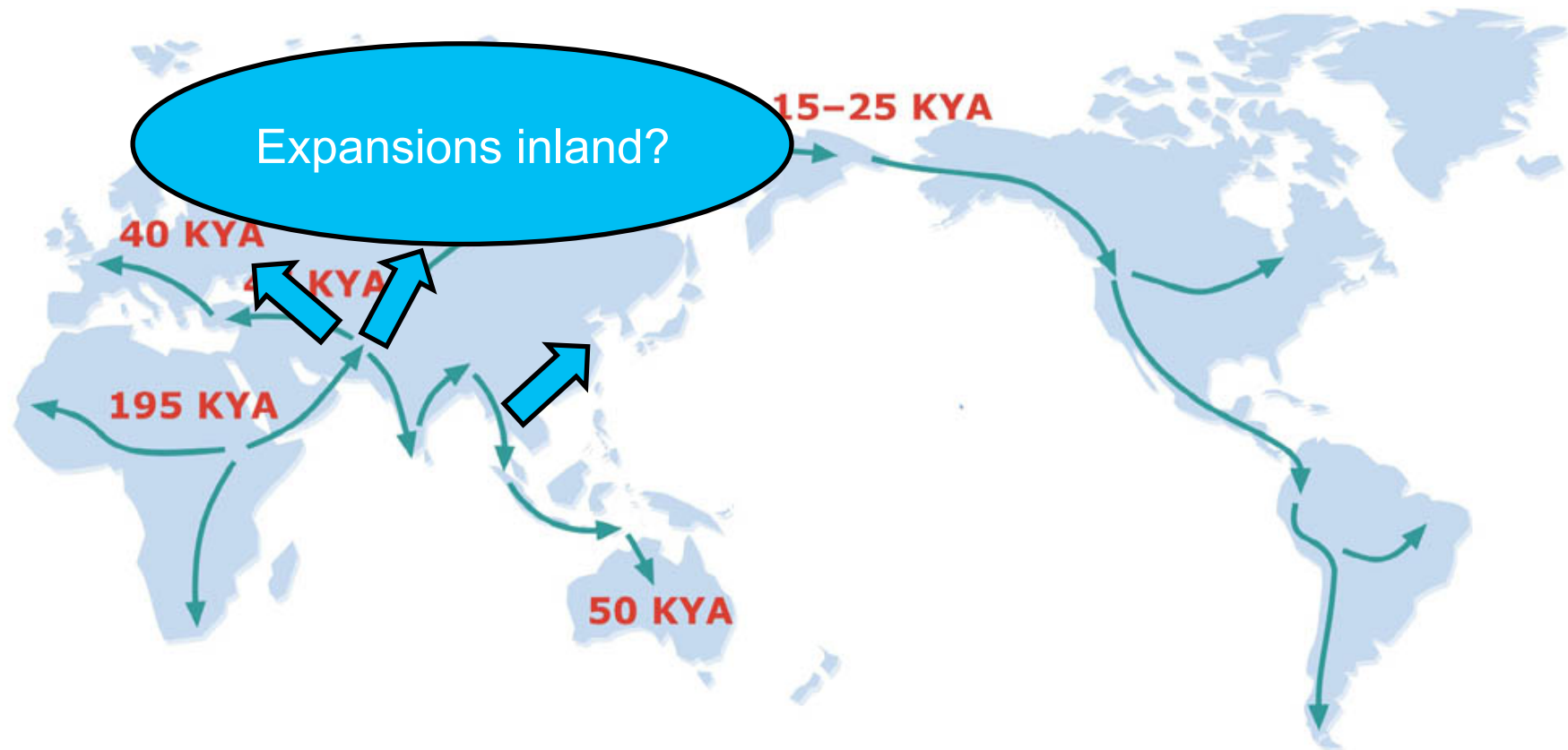
Out of Africa 55-75 KYA



What happened 40-50 KYA?

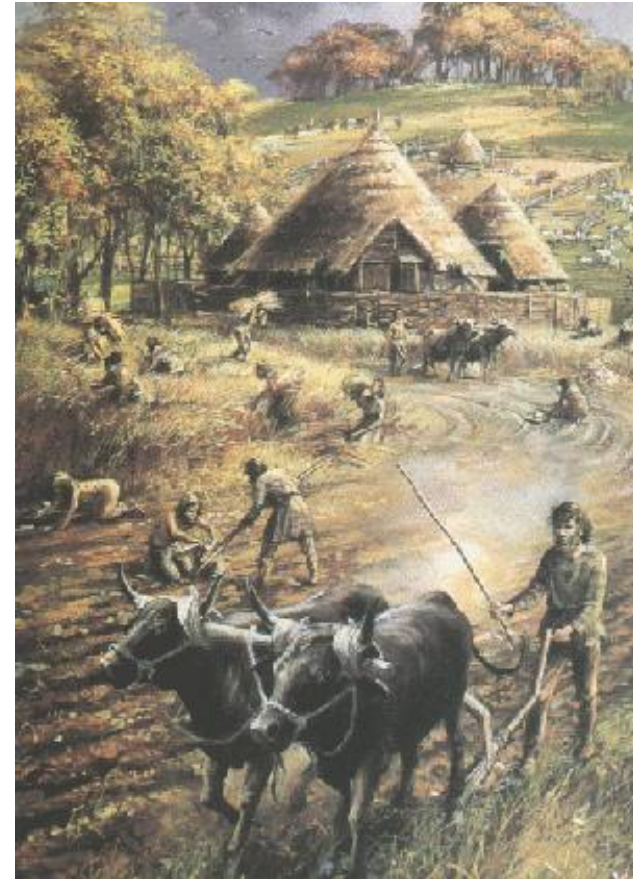


What happened 40-50 KYA?



R1b expansion in Europe

- Neolithic transition in Europe after ~10 KYA
- Does R1b represent a Paleolithic or Neolithic Y lineage?



Debate about R1b

Debate about R1b

The Genetic Legacy of Paleolithic *Homo sapiens* *sapiens* in Extant Europeans: A Y Chromosome Perspective

Ornella Semino,^{1,2*}† Giuseppe Passarino,^{2,3†} Peter J. Oefner,⁴
Alice A. Lin,² Svetlana Arbuzova,⁵ Lars E. Beckman,⁶
Giovanna De Benedictis,³ Paolo Francalacci,⁷
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Anna Mika,¹¹ Barbara Mika,¹² Dragan Primorac,¹³
A. Silvana Santachiara-Benerecetti,¹ L. Luca Cavalli-Sforza,²
Peter A. Underhill²

SCIENCE VOL 290 10 NOVEMBER 2000

marks the Aurignac settlement in Europe or,
at least, predates the LGM.

~41-47 KYA

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A Predominantly Neolithic Origin for European Paternal Lineages

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Rosser¹, Jane Goodwin², Jean-Paul Moisan³, Christelle Richard³, Ann Millward⁴, Andrew G. Demaine⁴,
Guido Barbujani⁵, Carlo Previderè⁶, Ian J. Wilson⁷, Chris Tyler-Smith⁸, Mark A. Jobling^{1*}

January 2010 | Volume 8 | Issue 1 | e1000285

6.5 (4.5-9.0) KYA

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The peopling of Europe and the cautionary tale of Y chromosome lineage R-M269

George B. J. Busby¹, Francesca Brisighelli^{1,3,4}, Paula Sánchez-Diz³,
Eva Ramos-Luis³, Conrado Martínez-Cadenas¹, Mark G. Thomas⁶,
Daniel G. Bradley⁷, Leonor Gusmão⁸, Bruce Winney²,
Walter Bodmer², Marielle Vennemann^{9,10}, Valentina Coia^{4,11},
Francesca Scarnicci¹², Sergio Tofanelli¹³, Giuseppe Vona¹⁴,
Rafal Ploski¹⁵, Carla Vecchiotti³, Tatijana Zemunik¹⁶, Igor Rudan^{16,17},
Sena Karachanak¹⁸, Draga Toncheva¹⁸, Paolo Anagnostou^{4,19},
Gianmarco Ferri²⁰, Cesare Rapone²¹, Tor Hervig²², Torolf Moen²³,
James F. Wilson^{17,24} and Cristian Capelli^{1,*}

Proc. R. Soc. B

doi:10.1098/rspb.2011.1044

existing data and tools are insufficient to make credible estimates :

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SCIENCE VOL 290 10 NOVEMBER 2000

marks the Aurignac settlement in Europe or, at least, predates the LGM.

~41-47 KYA

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PLOS BIOLOGY

A Predominantly Neolithic Origin for European Paternal Lineages

Patricia Balaresque¹, Georgina R. Bowden¹, Susan M. Adams¹, Ho-Yee Leung¹, Turi E. King¹, Zoë H. Rosser¹, Jane Goodwin², Jean-Paul Moisan³, Christelle Richard³, Ann Millward⁴, Andrew G. Demaine⁴, Guido Barbujani⁵, Carlo Previderè⁶, Ian J. Wilson⁷, Chris Tyler-Smith⁸, Mark A. Jobling^{1*}

January 2010 | Volume 8 | Issue 1 | e1000285

6.5 (4.5-9.0) KYA

The peopling of Europe and the cautionary tale of Y chromosome lineage R-M269

George B. J. Busby¹, Francesca Brisighelli^{1,3,4}, Paula Sánchez-Diz³, Eva Ramos-Luis³, Conrado Martínez-Cadenas¹, Mark G. Thomas⁶, Daniel G. Bradley⁷, Leonor Gusmão⁸, Bruce Winney², Walter Bodmer², Marielle Vennemann^{9,10}, Valentina Coia^{4,11}, Francesca Scarnicci¹², Sergio Tofanelli¹³, Giuseppe Vona¹⁴, Rafal Ploski¹⁵, Carla Vecchiotti³, Tatijana Zemunik¹⁶, Igor Rudan^{16,17}, Sena Karachanak¹⁸, Draga Toncheva¹⁸, Paolo Anagnostou^{4,19}, Gianmarco Ferri²⁰, Cesare Rapone²¹, Tor Hervig²², Torolf Moen²³, James F. Wilson^{17,24} and Cristian Capelli^{1,*}

Proc. R. Soc. B

doi:10.1098/rspb.2011.1044

existing data and tools are insufficient to make credible estimates

Debate about R1b

The Genetic Legacy of Paleolithic *Homo sapiens* in Extant Europeans: A Y Chromosome Perspective

Ornella Semino,^{1,2*} Giuseppe Passarino,^{2,3†} Peter J. Oefner,⁴ Alice A. Lin,² Svetlana Arbuzova,⁵ Lars E. Beckman,⁶ Giovanna De Benedictis,³ Paolo Francalacci,⁷ Anastasia Kouvatsi,⁸ Svetlana Limborska,⁹ Mladen Marcikiae,¹⁰ Anna Mika,¹¹ Barbara Mika,¹² Dragan Primorac,¹³ A. Silvana Santachiara-Benerecetti,¹ L. Luca Cavalli-Sforza,² Peter A. Underhill²

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Sequencing:
4-13 KYA

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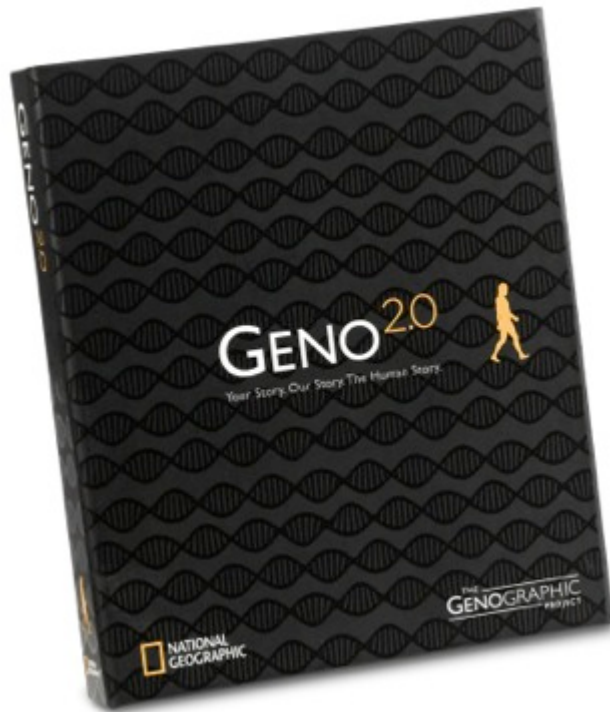
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Geno 2.0



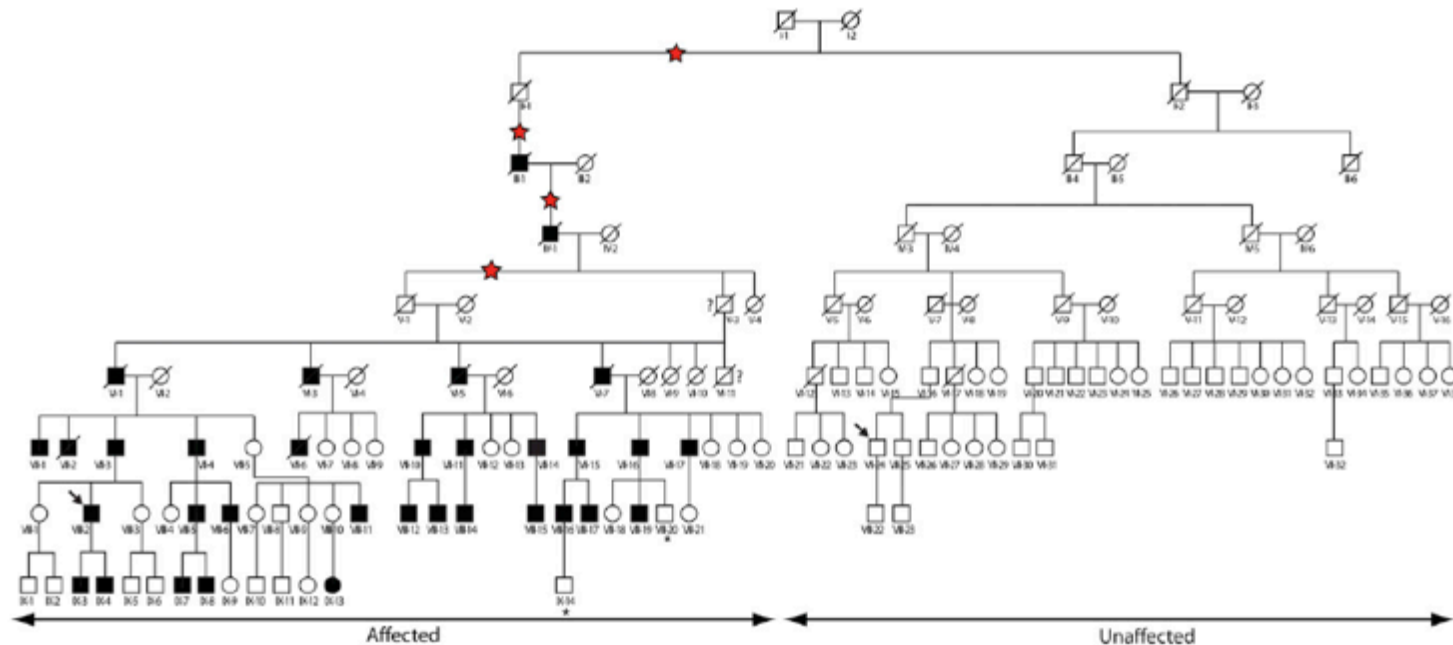
- ~150 K ancestry-informative, medically-uninformative SNPs
- ~15 K Y-SNPs
 - Karafet + published additions
 - FTDNA projects
 - 1000 Genomes Pilot
 - F: Li Jin, resequencing Chinese (3,753)
 - CTS: Chris Tyler-Smith, resequencing 1000 Genomes Phase 1 (3,850)
 - PF: Paolo Francalacci, resequencing Sardinians (5,400)

A mendelian disorder linked to the Y chromosome

REPORT

Genetic Basis of Y-Linked Hearing Impairment

Qiuju Wang,^{1,2} Yali Xue,² Yujun Zhang,^{2,5} Quan Long,^{2,6} Asan,^{2,3} Fengtang Yang,² Daniel J. Turner,^{2,7} Tomas Fitzgerald,² Bee Ling Ng,² Yali Zhao,¹ Yuan Chen,² Qingjie Liu,⁴ Weiyan Yang,¹ Dongyi Han,¹ Michael A. Quail,² Harold Swerdlow,² John Burton,² Ciara Fahey,² Zemin Ning,² Matthew E. Hurles,² Nigel P. Carter,² Huanming Yang,³ and Chris Tyler-Smith^{2,*}

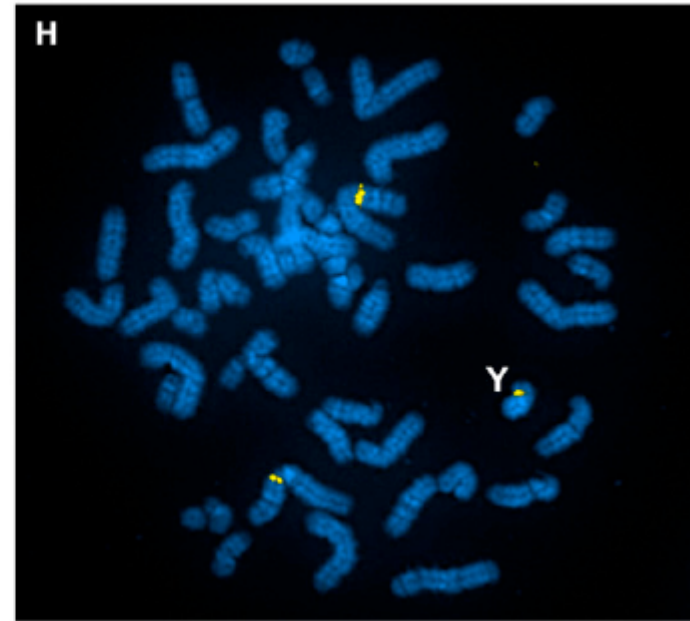
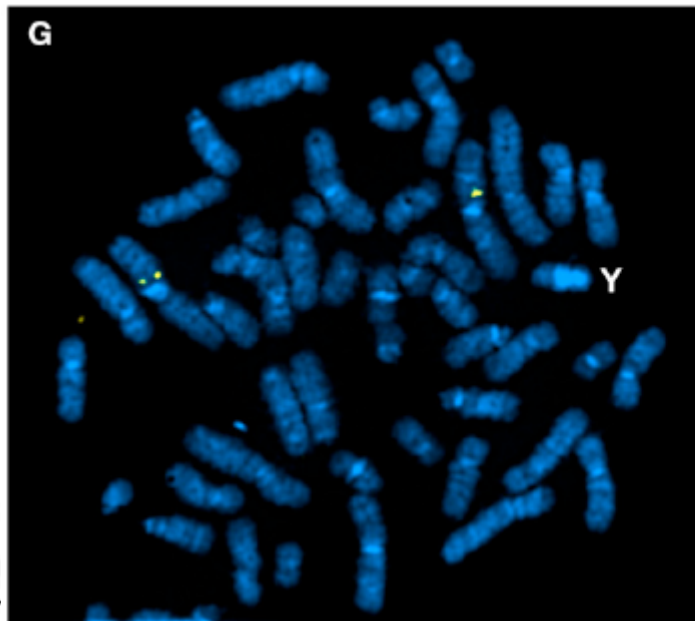


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REPORT

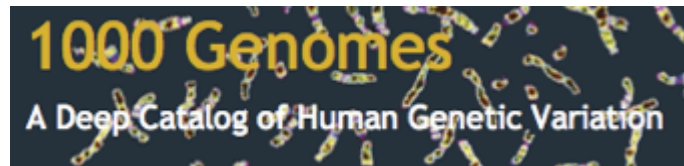
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Future projects and issues

Many large-scale genotyping and sequencing projects



People of the *British Isles*



SardiNIA

What Y reference sequence should we use?

- Current reference is a hybrid of hgs R1b and G
- Options:
 - Continue with this
 - All R1b
 - Inferred ancestral sequence

How should we name Y haplogroups?

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- R1b1b2g2b1a1a1b2a1b1a2c1a1b etc.?

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- R1b-14,057,355A
- Abbreviated names for major clusters only?
- Other?



Predictions for 2013

- Y-SNPs discovered from sequencing 1000s of individuals will be published
- There will be a need to compare and combine datasets
- Our current nomenclature system will break down
- More opportunities than ever for computer-literate citizen scientists

Acknowledgements



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